



dopamine beta-hydroxylase deficiency

Dopamine beta (β)-hydroxylase deficiency is a condition that affects the autonomic nervous system, which controls involuntary body processes such as the regulation of blood pressure and body temperature. Problems related to this disorder can first appear during infancy. Early signs and symptoms may include episodes of vomiting, dehydration, decreased blood pressure (hypotension), difficulty maintaining body temperature, and low blood sugar (hypoglycemia).

Individuals with dopamine β -hydroxylase deficiency typically experience a sharp drop in blood pressure upon standing (orthostatic hypotension), which can cause dizziness, blurred vision, or fainting. This sudden drop in blood pressure is usually more severe when getting out of bed in the morning, during hot weather, and as a person gets older. People with dopamine β -hydroxylase deficiency experience extreme fatigue during exercise (exercise intolerance) due to their problems maintaining a normal blood pressure.

Other features of dopamine β -hydroxylase deficiency include droopy eyelids (ptosis), nasal congestion, and an inability to stand for a prolonged period of time. Affected males may also experience retrograde ejaculation, a discharge of semen backwards into the bladder. Less common features include an unusually large range of joint movement (hypermobility) and muscle weakness.

Frequency

Dopamine β -hydroxylase deficiency is a very rare disorder. Fewer than 20 affected individuals, all of Western European descent, have been described in the scientific literature.

Genetic Changes

Mutations in the *DBH* gene cause dopamine β -hydroxylase deficiency. The *DBH* gene provides instructions for producing the enzyme dopamine β -hydroxylase. This enzyme converts dopamine to norepinephrine, both of which are chemical messengers (neurotransmitters) that transmit signals between nerve cells.

DBH gene mutations result in the production of a nonfunctional dopamine β -hydroxylase enzyme. People who lack functional dopamine β -hydroxylase cannot convert dopamine to norepinephrine, which leads to a shortage of norepinephrine in the

body. The lack of norepinephrine causes difficulty with regulating blood pressure and other autonomic nervous system problems seen in dopamine β -hydroxylase deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- dopamine β -hydroxylase
- noradrenaline deficiency
- norepinephrine deficiency

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Dopamine beta hydroxylase deficiency
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857209/>

Other Diagnosis and Management Resources

- GeneReview: Dopamine Beta-Hydroxylase Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK1474>
- Vanderbilt Autonomic Dysfunction Center
<https://ww2.mc.vanderbilt.edu/adx/4792>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Autonomic Nervous System Disorders
<https://medlineplus.gov/autonomicnervoussystemdisorders.html>

Genetic and Rare Diseases Information Center

- Dopamine beta hydroxylase deficiency
<https://rarediseases.info.nih.gov/diseases/1903/dopamine-beta-hydroxylase-deficiency>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Dysautonomia Information Page
<https://www.ninds.nih.gov/Disorders/All-Disorders/Dysautonomia-Information-Page>

Educational Resources

- Cleveland Clinic: Dysautonomia
<http://my.clevelandclinic.org/health/articles/dysautonomia>
- Disease InfoSearch: Dopamine Beta Hydroxylase Deficiency
<http://www.diseaseinfosearch.org/Dopamine+Beta+Hydroxylase+Deficiency/2316>
- MalaCards: dopamine beta-hydroxylase deficiency
http://www.malacards.org/card/dopamine_beta_hydroxylase_deficiency
- Merck Manual Consumer Version: Autonomic Neuropathies
<http://www.merckmanuals.com/home/brain-spinal-cord-and-nerve-disorders/autonomic-nervous-system-disorders/autonomic-neuropathies>
- Merck Manual Consumer Version: Dizziness or Light-Headedness When Standing Up
<http://www.merckmanuals.com/home/heart-and-blood-vessel-disorders/symptoms-of-heart-and-blood-vessel-disorders/dizziness-or-light-headedness-when-standing-up>
- Orphanet: Dopamine beta-hydroxylase deficiency
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=230
- Washington University, St. Louis: Neuromuscular Disease Center
<http://neuromuscular.wustl.edu/autonomic.html#dbh>

Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases
<http://www.climb.org.uk/>

GeneReviews

- Dopamine Beta-Hydroxylase Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK1474>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22dopamine+beta-hydroxylase+deficiency%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28dopamine+beta-hydroxylase+deficiency%5BTIAB%5D%29+OR+%28norepinephrine+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL
<http://omim.org/entry/223360>

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